



VPS13A gene

vacuolar protein sorting 13 homolog A

Normal Function

The *VPS13A* gene provides instructions for producing a protein called chorein. Chorein is found in various tissues throughout the body. The function of this protein is unknown. Some researchers believe that chorein plays a role in the movement of proteins within cells.

Health Conditions Related to Genetic Changes

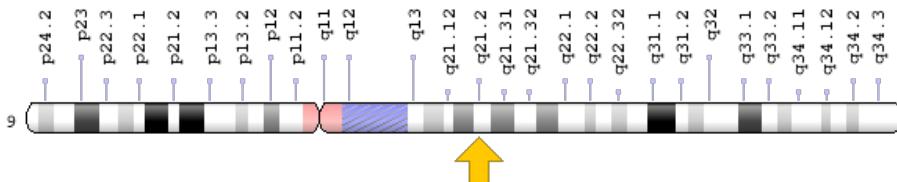
chorea-acanthocytosis

Approximately 75 mutations in the *VPS13A* gene have been found to cause chorea-acanthocytosis. Most of these mutations insert or delete one or more building blocks of DNA (nucleotides) in the *VPS13A* gene. These changes in the gene lead to the production of an abnormally small, nonfunctional version of chorein. Many of the mutations are specific to single families, although people with chorea-acanthocytosis who are French-Canadian or Japanese tend to have a mutation that is specific to their population. It is unclear why mutations in the *VPS13A* gene affect only the brain and red blood cells, causing the signs and symptoms of chorea-acanthocytosis.

Chromosomal Location

Cytogenetic Location: 9q21.2, which is the long (q) arm of chromosome 9 at position 21.2

Molecular Location: base pairs 77,177,353 to 77,421,537 on chromosome 9 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CHAC
- CHOREIN
- FLJ42030
- KIAA0986
- vacuolar protein sorting 13 homolog A (*S. cerevisiae*)
- vacuolar protein sorting 13A
- VP13A_HUMAN

Additional Information & Resources

GeneReviews

- Chorea-Acanthocytosis
<https://www.ncbi.nlm.nih.gov/books/NBK1387>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28VPS13A%5BTIAB%5D%29+OR+%28chorein%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2160+days%22%5Bdp%5D>

OMIM

- VACUOLAR PROTEIN SORTING 13, YEAST, HOMOLOG OF, A
<http://omim.org/entry/605978>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_VPS13A.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=VPS13A%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=1908
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/23230>
- UniProt
<http://www.uniprot.org/uniprot/Q96RL7>

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